



WNK4 gene

WNK lysine deficient protein kinase 4

Normal Function

The *WNK4* gene provides instructions for making a protein that plays a role in blood pressure regulation by helping control the amount of sodium and potassium in the body. The WNK4 protein acts as a kinase, which is an enzyme that changes the activity of other proteins by adding a cluster of oxygen and phosphorus atoms (a phosphate group) at specific positions.

The WNK4 protein regulates channels in the cell membrane that control the transport of sodium or potassium into and out of cells, which occurs primarily in the kidneys. Sodium channels help transport sodium into specialized kidney cells, which then transfer it into the blood. This transfer helps keep sodium in the body through a process called reabsorption. Potassium channels handle excess potassium that has been transferred from the blood into the kidney cells. The channels transport the potassium out of the cells in a process called secretion, so that it can be removed from the body in the urine. The WNK4 protein is able to promote sodium reabsorption and block potassium secretion. Depending on conditions in the cell, the WNK4 protein is also able to block (inhibit) sodium reabsorption.

Health Conditions Related to Genetic Changes

pseudohypoaldosteronism type 2

At least eight mutations in the *WNK4* gene have been found to cause pseudohypoaldosteronism type 2 (PHA2), a condition characterized by high blood pressure (hypertension) and high levels of potassium in the blood (hyperkalemia). The mutations involved in this condition change single protein building blocks (amino acids) in the WNK4 protein. The alterations to the WNK4 protein impair its breakdown, resulting in higher than normal levels of WNK4 protein and elevated WNK4 activity. The increase in WNK4 activity leads to increased sodium reabsorption and reduced potassium secretion, resulting in hypertension and hyperkalemia.

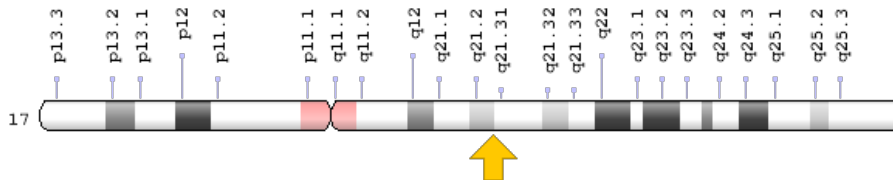
other disorders

Studies have associated normal variations in the *WNK4* gene with an increased risk of high blood pressure (hypertension) in people without pseudohypoaldosteronism type 2. A combination of genetic variations and environmental factors likely influence the development of this complex condition.

Chromosomal Location

Cytogenetic Location: 17q21.2, which is the long (q) arm of chromosome 17 at position 21.2

Molecular Location: base pairs 42,779,701 to 42,797,066 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- PHA2B
- PRKWNK4
- protein kinase lysine-deficient 4
- protein kinase with no lysine 4
- serine/threonine-protein kinase WNK4
- WNK4_HUMAN

Additional Information & Resources

GeneReviews

- Pseudohypoaldosteronism Type II
<https://www.ncbi.nlm.nih.gov/books/NBK65707>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28WNK4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- PROTEIN KINASE, LYSINE-DEFICIENT 4
<http://omim.org/entry/601844>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_WNK4.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=WNK4%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14544
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/65266>
- UniProt
<http://www.uniprot.org/uniprot/Q96J92>

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